Abstracts *(Oral Presentation)*

Acute Intermittent Porphyria: Female Adolescent with Complete Phenotypes in p.Arg173Trp Variant in Thailand

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Abstract

Introduction: Acute intermittent porphyria (AIP) is a rare disease with a prevalence of around 1:200,000 symptomatic AIP, caused by the deficiency of porphobilinogen deaminase enzyme, in heme synthesis pathway. Heterozygous pathogenic variants in *HMBS* is associated with AIP. Multisystemic manifestation of acute neurovisceral features are quite challenging for diagnosis. Currently, less than five patients have been reported on AIP in Thailand, therefore, it remains difficult to infer whether the prevalence in Thai people is less than other countries or that the diagnosis of AIP is not made.

Objectives: To review clinical course of patient who had a definite diagnosis of AIP, and to perform the genetic testing in all available relatives.

Methods: All available relatives of AIP patient were enrolled and the genetic test was performed for pathogenic variant of *HMBS*.

Results: Our patient, 14-year-old female, presented with severe abdominal pain, vomiting, seizure, posterior reversible encephalopathy syndrome syndrome of inappropriate antidiuretic hormone and muscle weakness, which are all classic phenotypes of AIP attack has a heterozygous pathogenic variant of *HMBS* at the codon 517; c.517C > T (p.Arg 173Trp). Genetic test revealed three in five participants carry this pathogenic variant; currently, all are latent AIP, but attack may occur.

Conclusions: This study would like to raise the awareness of the medical team, as your patient may have a rare disease. If you can give an earlier diagnosis, leads to prompt and specific treatment which can shorten the duration of attacks, prevent any complications, reduce the treatment cost, and reduce mortality rate.

Keywords: Acute intermittent porphyria, Porphobilinogen deaminase enzyme deficiency, *HMBS*
DOI: https://doi.org/10.14456/2022s10722

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